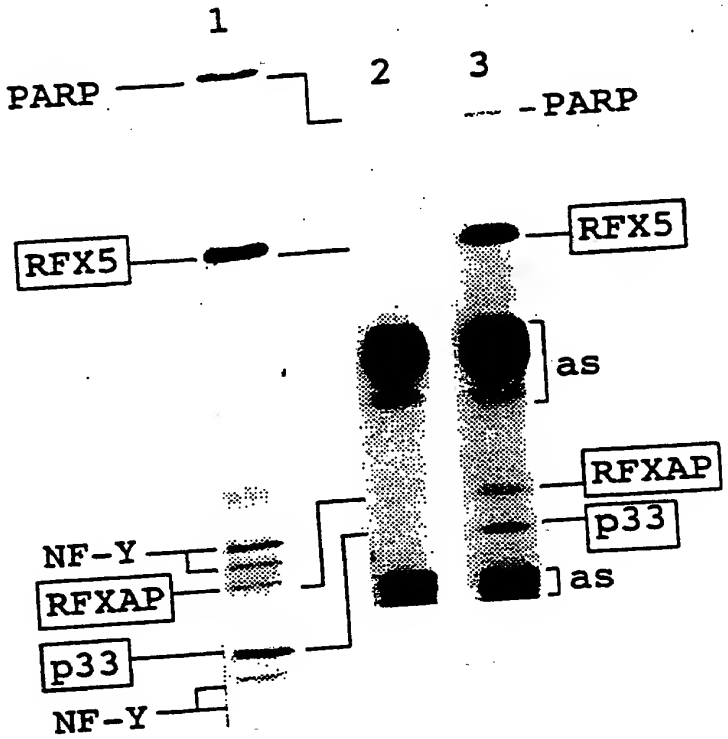


Figure 1

WO 2004/046340 PCT/EP2003/012940

```
                  ⟶ 1
1       ACGCAGGGAAGGAGGCACACCCGGGGGTGGCGCAGTGAGGAGGGGGCGCGACGGCCA

58      GGAGGCTGGTGGAGCGACACCCAGGCAGGAGAGGGGGAAGAACTCTCTCCCTTTCTGAAC

118     CCCCTTTTCCTTGAGAGACGAGTTGGGGGAGTCCTCCACGCATTACCCACTCGGGCCGCA

178     AAAACTCCCTTCTTTAGCCCTCTGCCCCCGCCCTTGCTTATAAGCCTTTGAGACCGCAGA

                                  ⟶ 2
238     AGGGACCTTGTTGTGGAACGGGACGGCCAAGAGGAAGCCAGATCGCTGAGGGTCCGGTCT

298     CCAGTTTGCCTCCTGCTATATCCATTGGAAGAGAAAAGTTTGTGACTTGGGCCCCCAAGT
                                                        ⟶ 3
358     TTTGAGAGAACTGGGCTTTCGGCGCGGGGGGACAGAGGAGGCTCGTGGGGAGCTTTCCCC

418     ATGGAGCTTACCCAGCCTGCAGAAGACCTCATCCAGACCCAGCAGACCCCTGCCTCAGAA
1        M  E  L  T  Q  P  A  E  D  L  I  Q  T  Q  Q  T  P  A  S  E

478     CTTGGGGACCCTGAAGACCCCGGAGAGGAGGCTGCAGATGGCTCAGACACTGTGGTCCTC
21       L  G  D  P  E  D  P  G  E  E  A  A  D  G  S  D  T  V  V  L

538     AGTCTCTTTCCCTGCACCCCTGAGCCTGTGAATCCTGAACCGGATGCCAGTGTTTCCTCT
41       S  L  F  P  C  T  P  E  P  V  N  P  E  P  D  A  S  V  S  S
                     ⟶ 4
598     CCACAGGcagGCAGCTCCCTGAAGCACTCCACCACTCTCACCAACCGGCAGCGAGGGAAC
61       P  Q  (A)  G  S  S  L  K  H  S  T  T  L  T  N  R  Q  R  G  N
```

Figure 2a

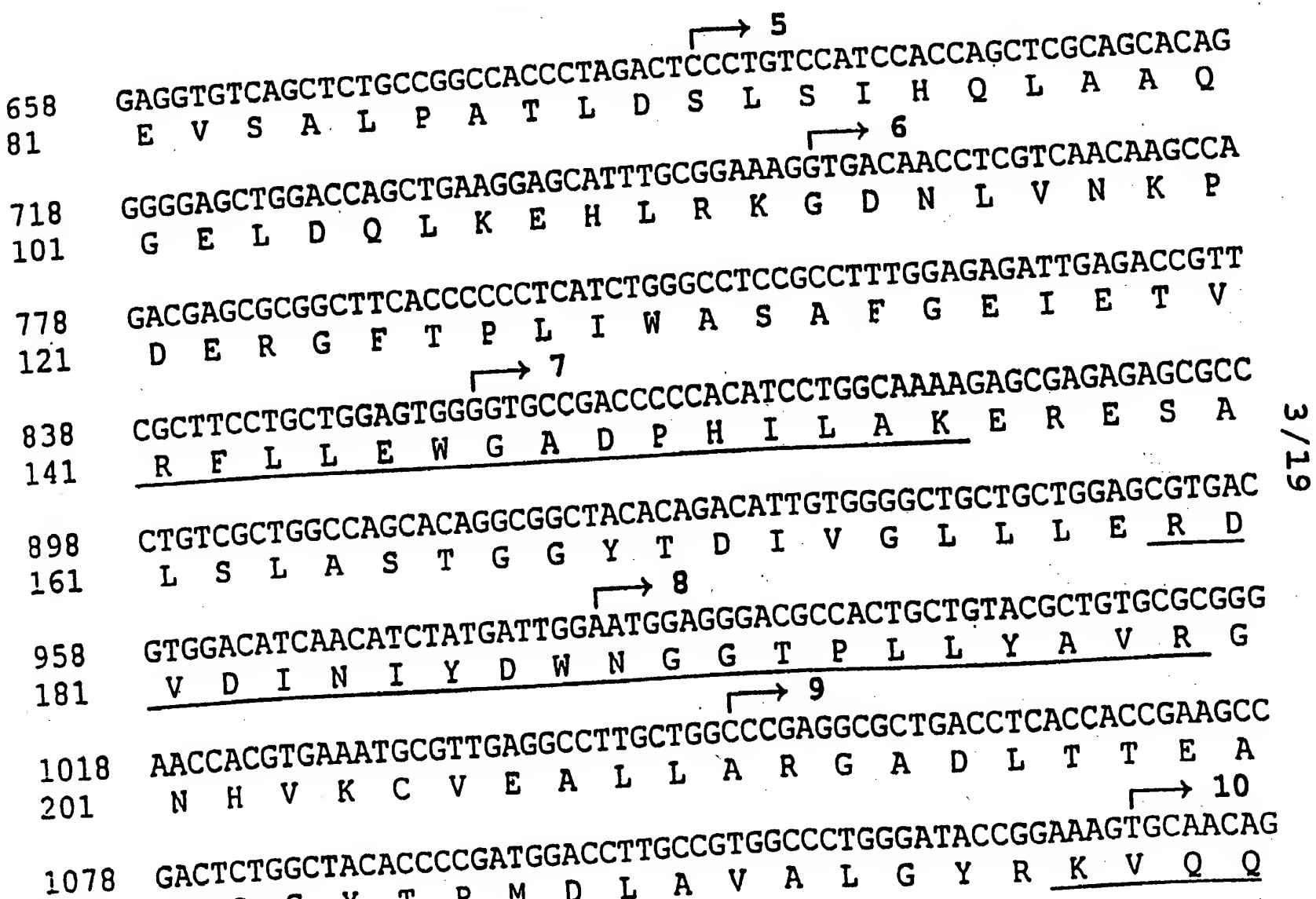

5
658 GAGGTGTCAGCTCTGCCGGCCACCCTAGACTCCCTGTCCATCCACCAGCTCGCAGCACAG
81 E V S A L P A T L D S L S I H Q L A A Q
6
718 GGGGAGCTGGACCAGCTGAAGGAGCATTTGCGGAAAGGTGACAACCTCGTCAACAAGCCA
101 G E L D Q L K E H L R K G D N L V N K P
778 GACGAGCGCGGCTTCACCCCCCTCATCTGGGCCTCCGCCTTTGGAGAGATTGAGACCGTT
121 D E R G F T P L I W A S A F G E I E T V
7
838 CGCTTCCTGCTGGAGTGGGGTGCCGACCCCCACATCCTGGCAAAAGAGCGAGAGAGCGCC
141 R F L L E W G A D P H I L A K E R E S A
898 CTGTCGCTGGCCAGCACAGGCGGCTACACAGACATTGTGGGGCTGCTGCTGGAGCGTGAC
161 L S L A S T G G Y T D I V G L L L E R D
8
958 GTGGACATCAACATCTATGATTGGAATGGAGGGACGCCACTGCTGTACGCTGTGCGCGGG
181 V D I N I Y D W N G G T P L L Y A V R G
9
1018 AACCACGTGAAATGCGTTGAGGCCTTGCTGGCCCGAGGCGCTGACCTCACCACCGAAGCC
201 N H V K C V E A L L A R G A D L T T E A
10
1078 GACTCTGGCTACACCCCGATGGACCTTGCCGTGGCCCTGGGATACCGGAAAGTGCAACAG
221 D S G Y T P M D L A V A L G Y R K V Q Q

Figure 2b

FOREIGN EXCHANGE

```
1138  GTGATCGAGAACCACATCCTCAAGCTCTTCCAGAGCAACCTGGTGCCCGCTGACCCTGAG
241    V  I  E  N  H  I  L  K  L  F  Q  S  N  L  V  P  A  D  P  E

1198  TGAAGGCCGCCTGCCGGGGACTCAGACACTCAGGGAACAAAATGGTCAGCCAGAGCTGGG
260    *

1258  GAAACCCAGAACTGACTTCAAAGGCAGCTTCTGGACAGGTGGTGGGAGGGGACCCTTCCC

1318  AAGAGGAACCAATAAACCTTCTGTGCAG - polyA
```


Figure 2c

Found 2m20m860

```
sRFXANK  MELTQPAEDLIQTQQTPASELGDPEDPGEEAADGSDTVVLSLFPCTPEPVNPEPDASVSS  60
mRFXANK  --P--V--N-VPN--P-VPD-E----TRD-SPEN-------------DA----A---A--  60
s homol                                NAFNVFTFVFHLAECNIHTSPSPGIQVRHVXTP-T  35
m homol                                ASVLFKAECNIHTSPSPGIQVRHVYTP-T  29

                                                HHHHT HHHHHHHHHHT
sRFXANK  PQaGSSLKHSTTLTNRQRGNEVSALPATLDSLSIHQLAAQGELDQLKEHLRKG..... 113
mRFXANK  L-.--F-----------------------------------S---D-----acpactc 119
s homol  TKHF-PI-Q------KH------TT-LLAN---V--------MLY-ATRIEQ.....  87
m homol  TKHF-PI-Q------KH------TT-LLAN---A--------MLY-ATRIEQ.....  81
ABPβ                                    EA-RA-QD-EVRILMAN-.....  25

                                  ank 1
                        TT   HHHHHHHHT HHHHHHHHTTT  TT   TT   HHHHHHHHT H
sRFXANK  ...DNLVNKPDERGFTPLIWASAFGEIETVRFLLEWGADPHILAKERESALSLASTGGYT 170
mRFXANK  lsgN--I-------------------------D-------------------M----- 179
s homol  ...E-VI-HT--E-----M--A-H-Q-AV-E---QN----QL-G-G--------CSK--- 144
m homol  ...E-VI-HT--E-----M--A-H-Q-AV-E---QN----QL-G-G--------CSK--- 138
ABPβ     ...APFTT..-WL-TS--HL-AQY-HFS-TEV--RA-VSRDART-VDRTP-HM-ASE-HA  80
```

Figure 3a

PCT/EP2004/050360

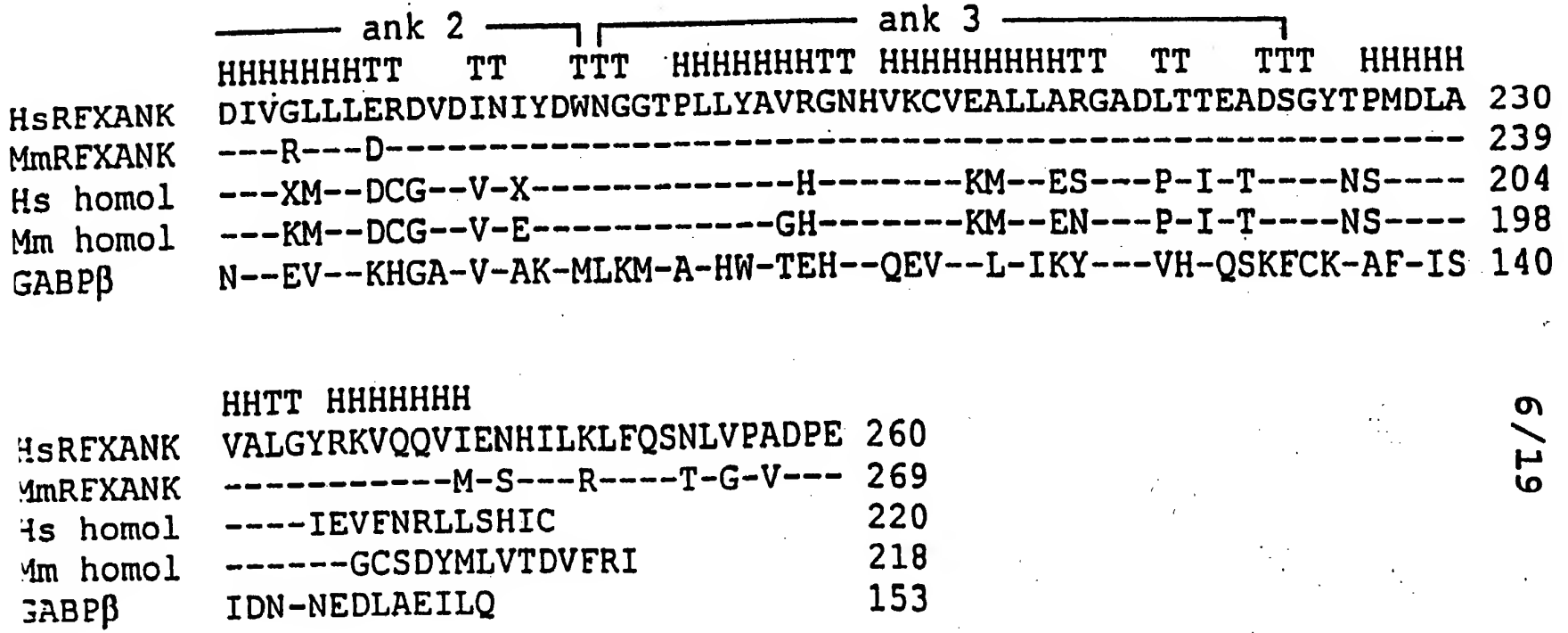


Figure 3b

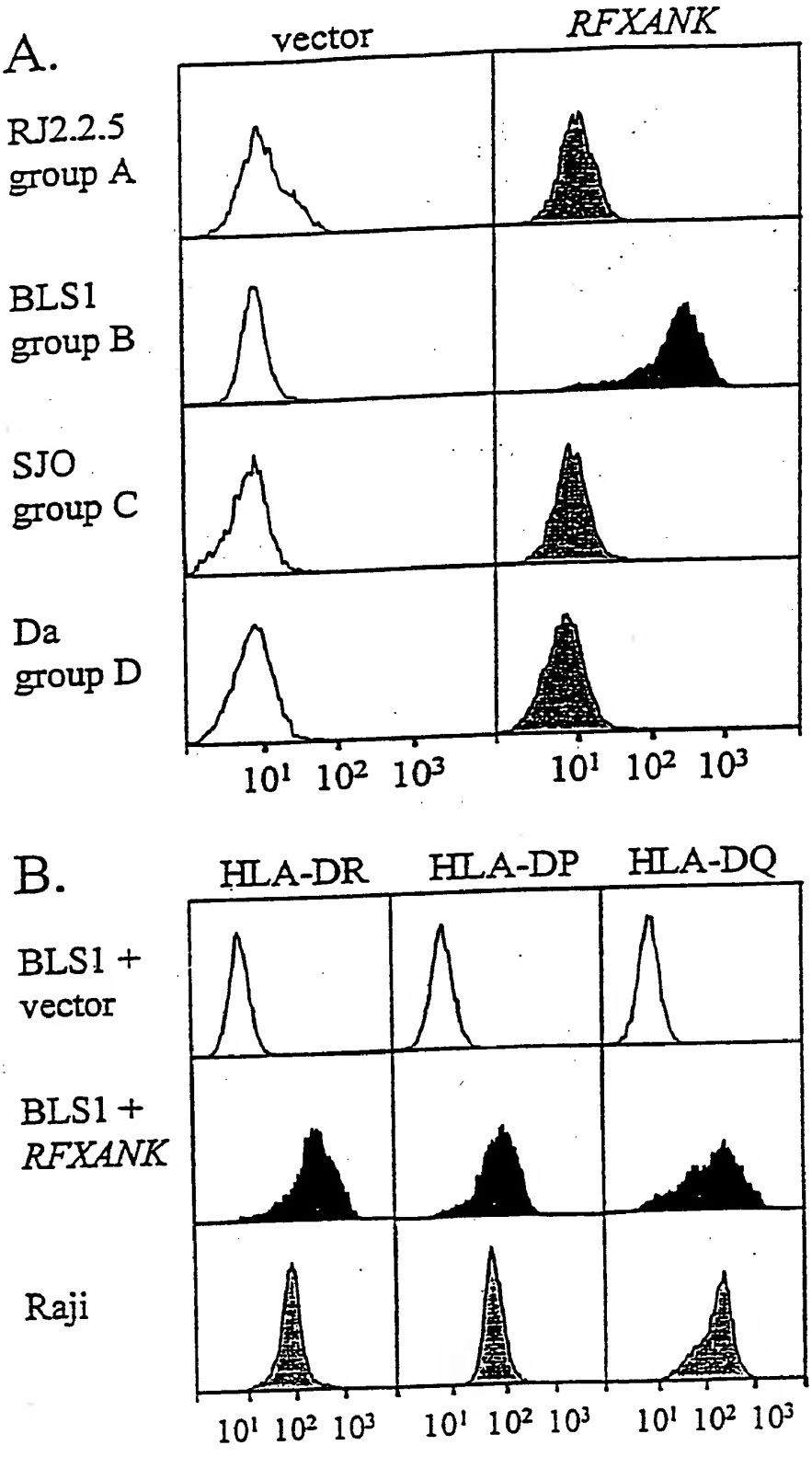
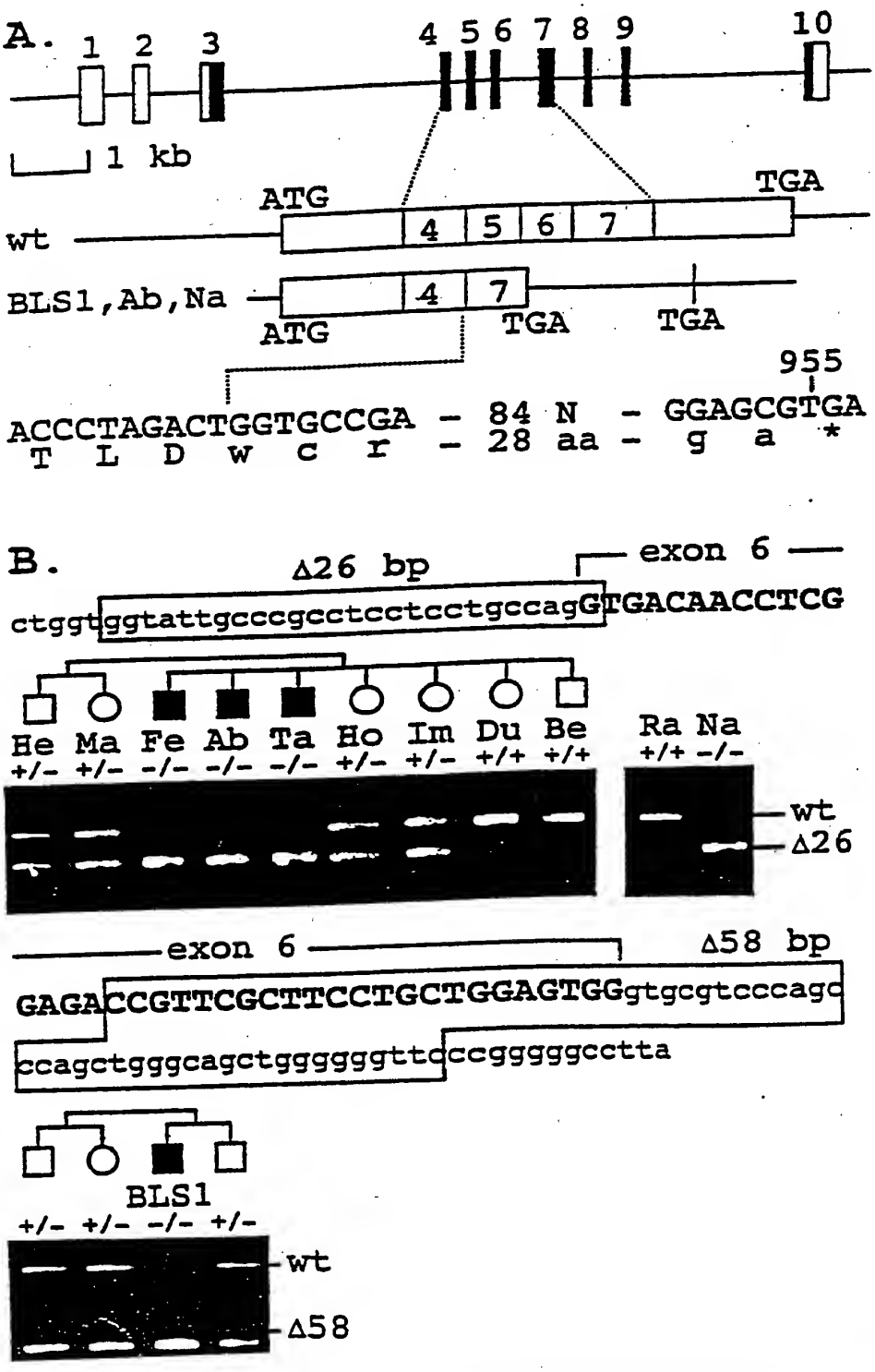

A.
vector
RFXANK
RJ2.2.5
group A
BLS1
group B
SJO
group C
Da
group D
10¹ 10² 10³
10¹ 10² 10³
B.
HLA-DR
HLA-DP
HLA-DQ
BLS1 +
vector
BLS1 +
RFXANK
Raji
10¹ 10² 10³
10¹ 10² 10³
10¹ 10² 10³

Figure 4


A.
1 2 3 4 5 6 7 8 9 10
1 kb
ATG
TGA
wt
4 5 6 7
BLS1,Ab,Na
4 7
ATG
TGA
TGA
955
ACCCTAGACTGGTGCCGA - 84 N - GGAGCGTGA
T L D w c r - 28 aa - g a *
B.
Δ26 bp
exon 6
ctggtggtattgcccgcctcctcctgccagGTGACAACCTCG
He Ma Fe Ab Ta Ho Im Du Be
+/- +/- -/- -/- -/- +/- +/- +/+ +/+
Ra Na
+/+ -/-
wt
Δ26
exon 6
Δ58 bp
GAGACCGTTCGCTTCCTGCTGGAGTGGgtgcgtcccagc
ccagctgggcagctgggggttcccgggggccttа
BLS1
+/- +/- -/- +/-
wt
Δ58

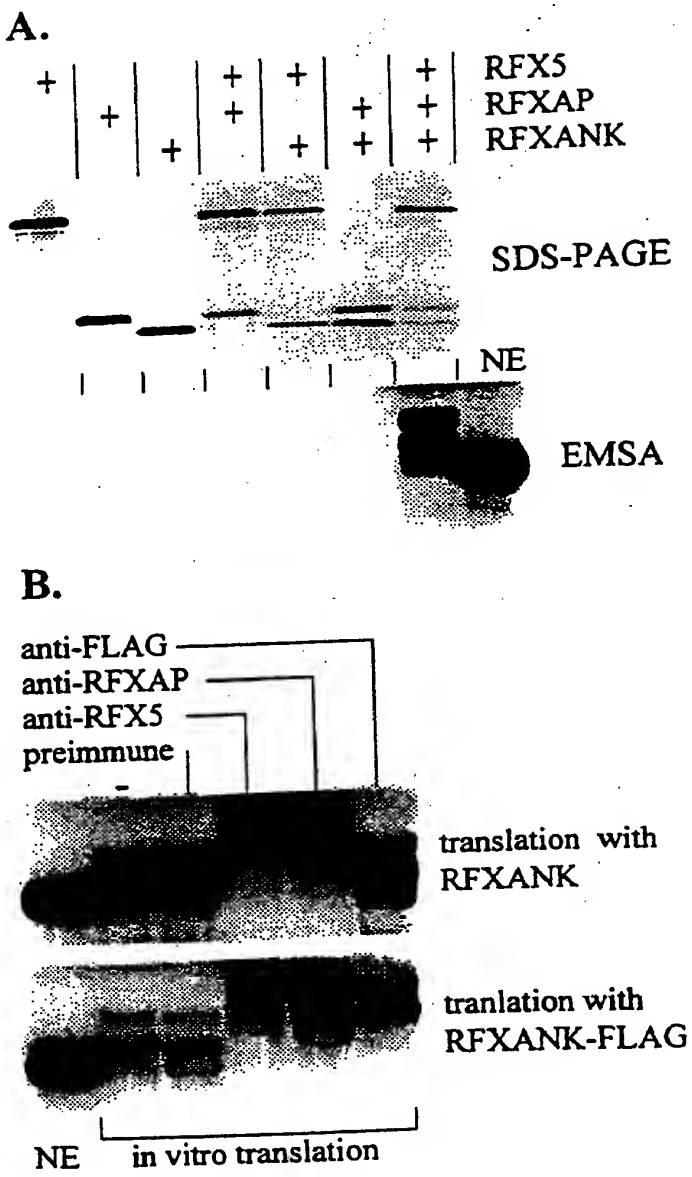

A.
RFX5
RFXAP
RFXANK
SDS-PAGE
NE
EMSA
B.
anti-FLAG
anti-RFXAP
anti-RFX5
preimmune
translation with RFXANK
tranlation with RFXANK-FLAG
NE
in vitro translation

Figure 6

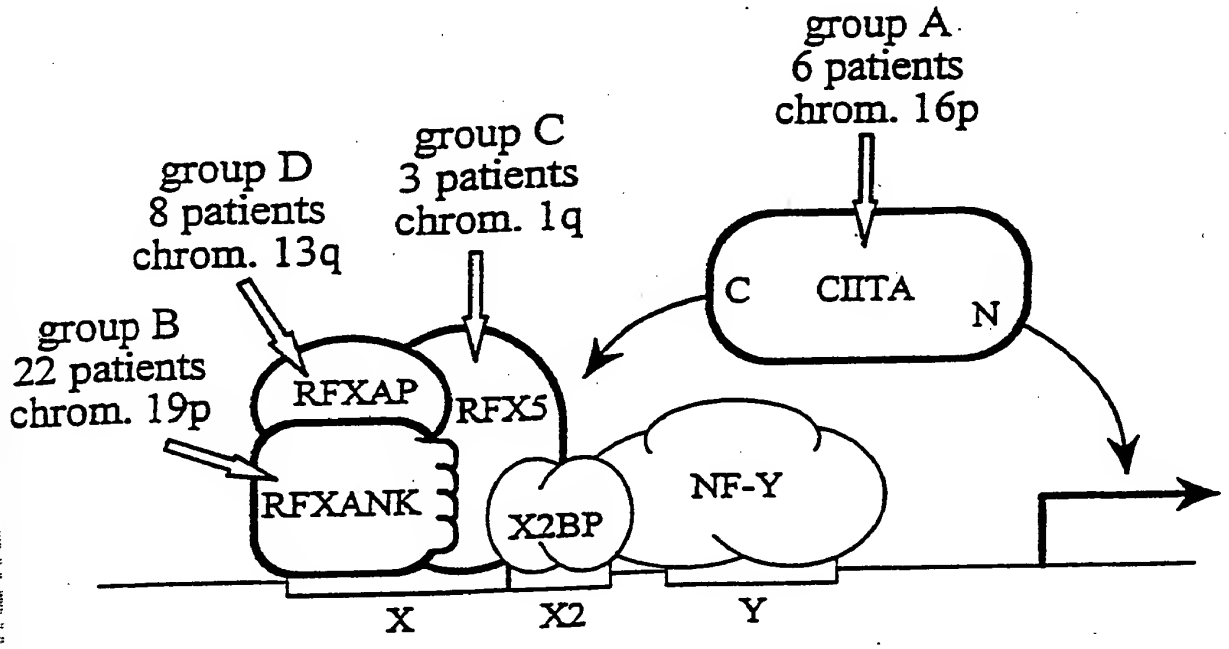

group A
6 patients
chrom. 16p
group C
3 patients
chrom. 1q
group D
8 patients
chrom. 13q
group B
22 patients
chrom. 19p
C
CIITA
N
RFXAP
RFX5
RFXANK
X2BP
NF-Y
X
X2
Y

Figure 7

PUBLISHED ENGLISH

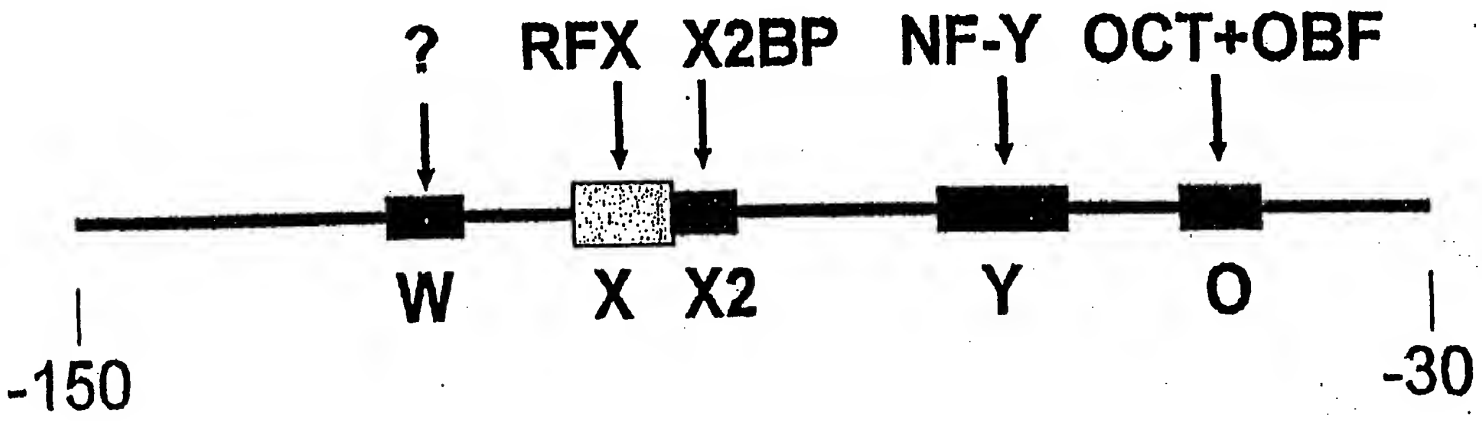


Figure 8

Found Medline

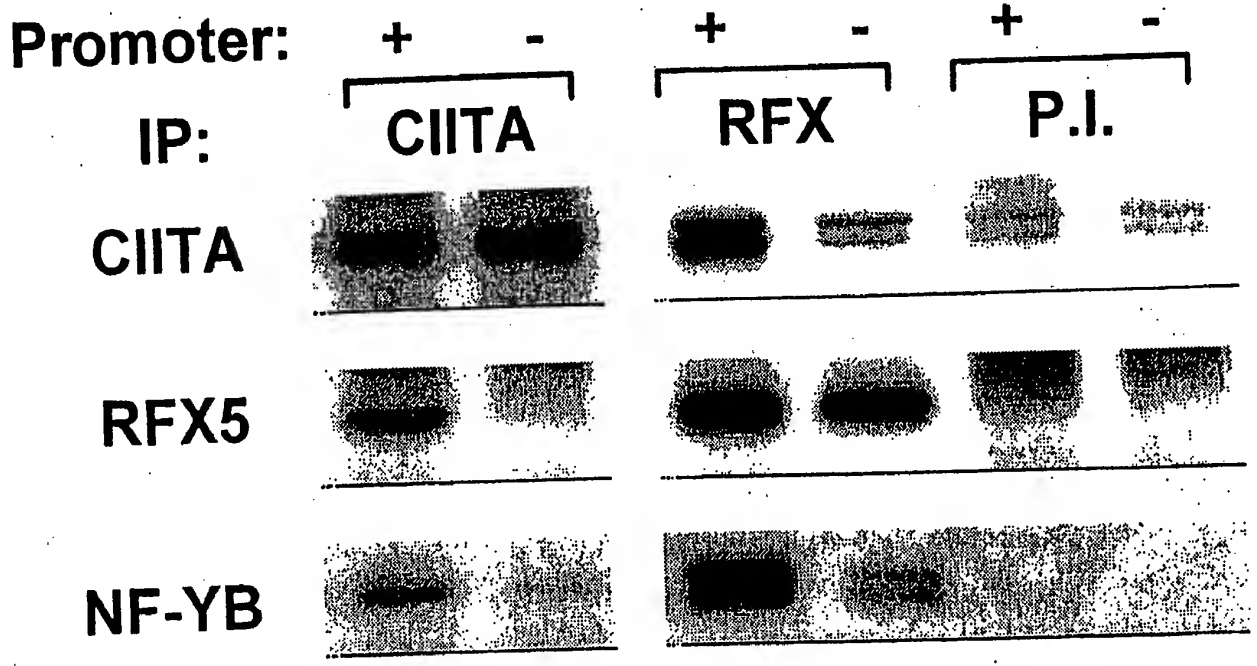


Figure 9

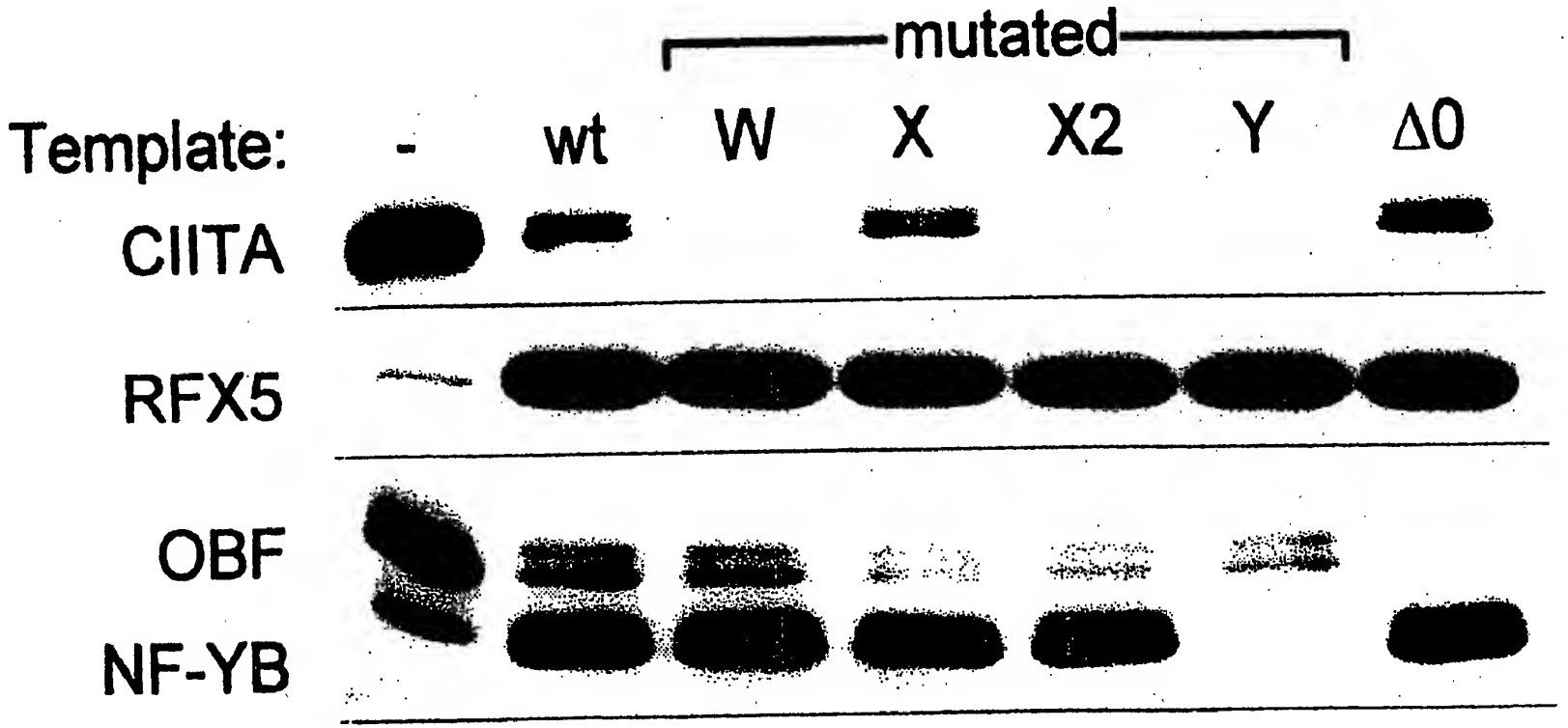


Figure 10a

Figure 10b

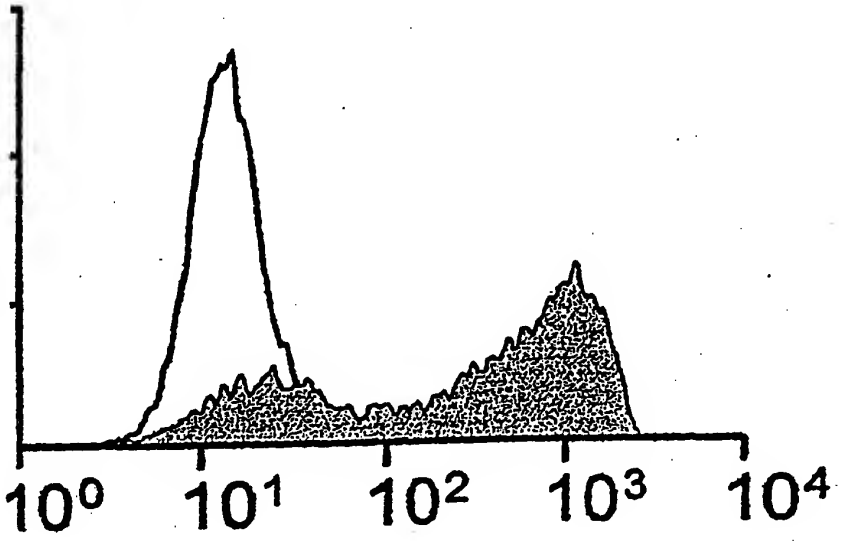


Figure 10c

Tomio Enomoto

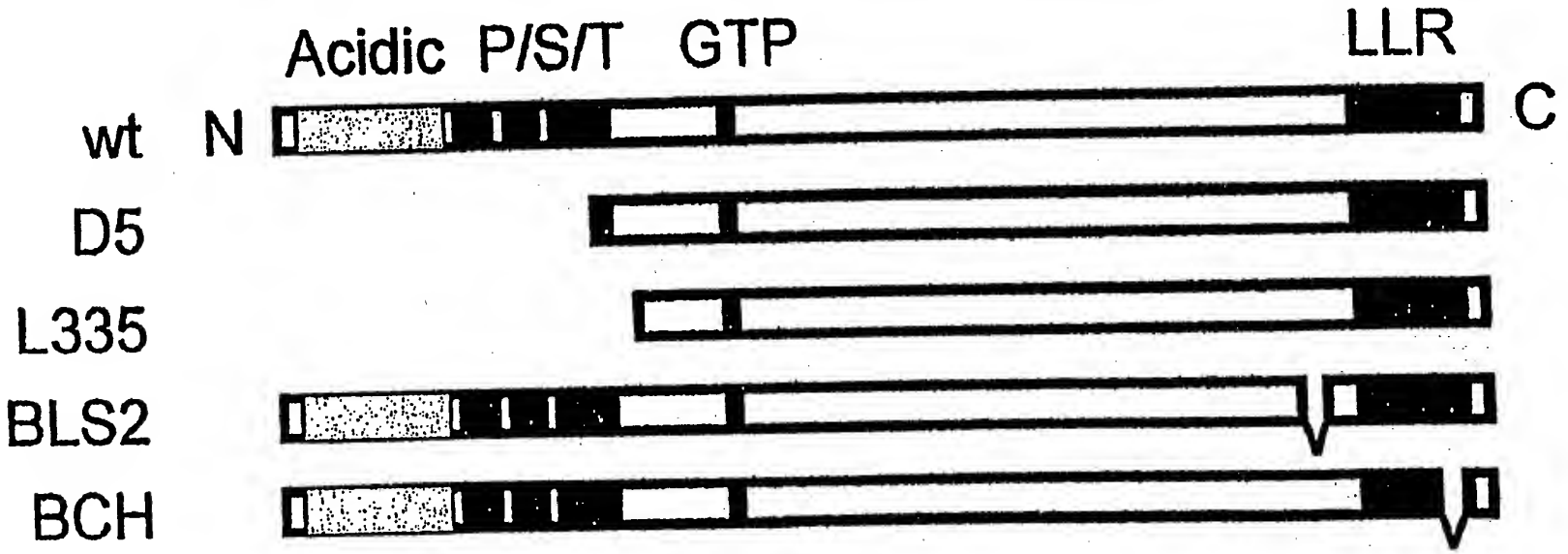


Figure 11a

ST/OT

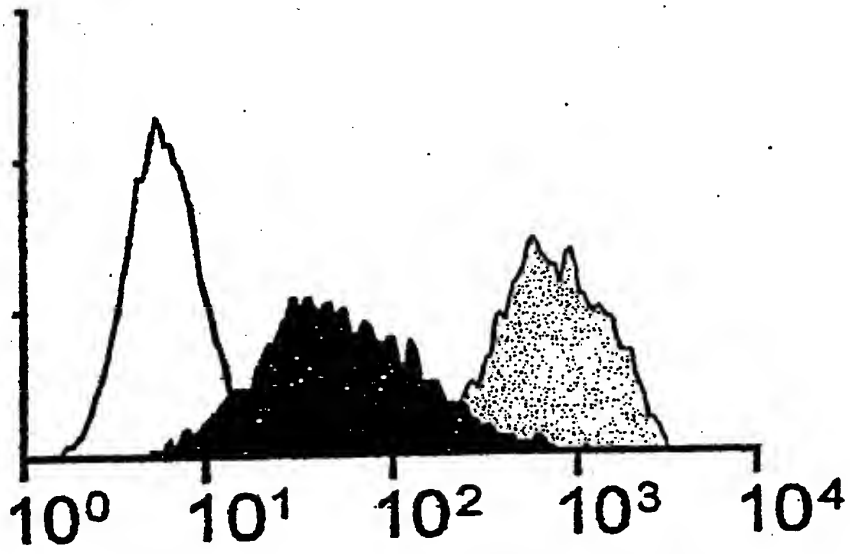


Figure 11b

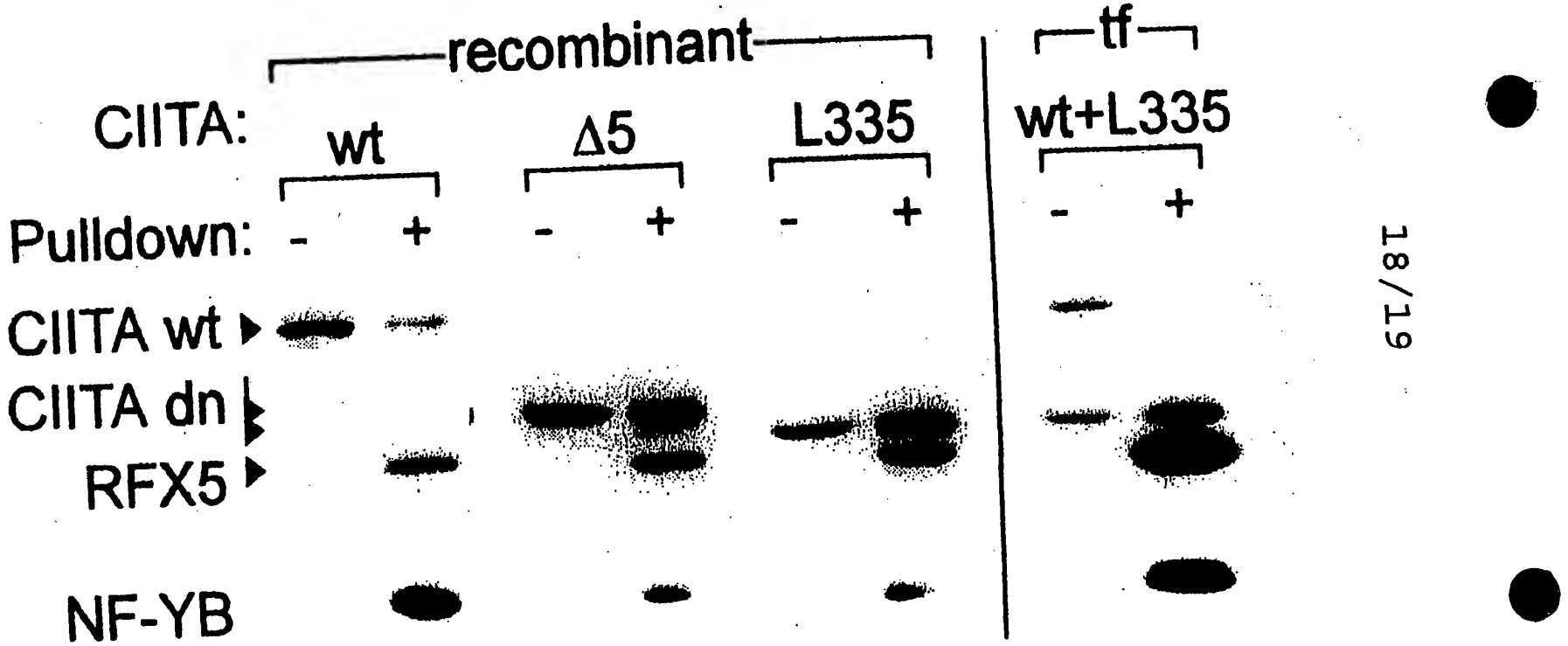

recombinant
tf
CIITA:
wt
Δ5
L335
wt+L335
Pulldown:
-
+
-
+
-
+
-
+
CIITA wt
CIITA dn
RFX5
NF-YB

Figure 11c

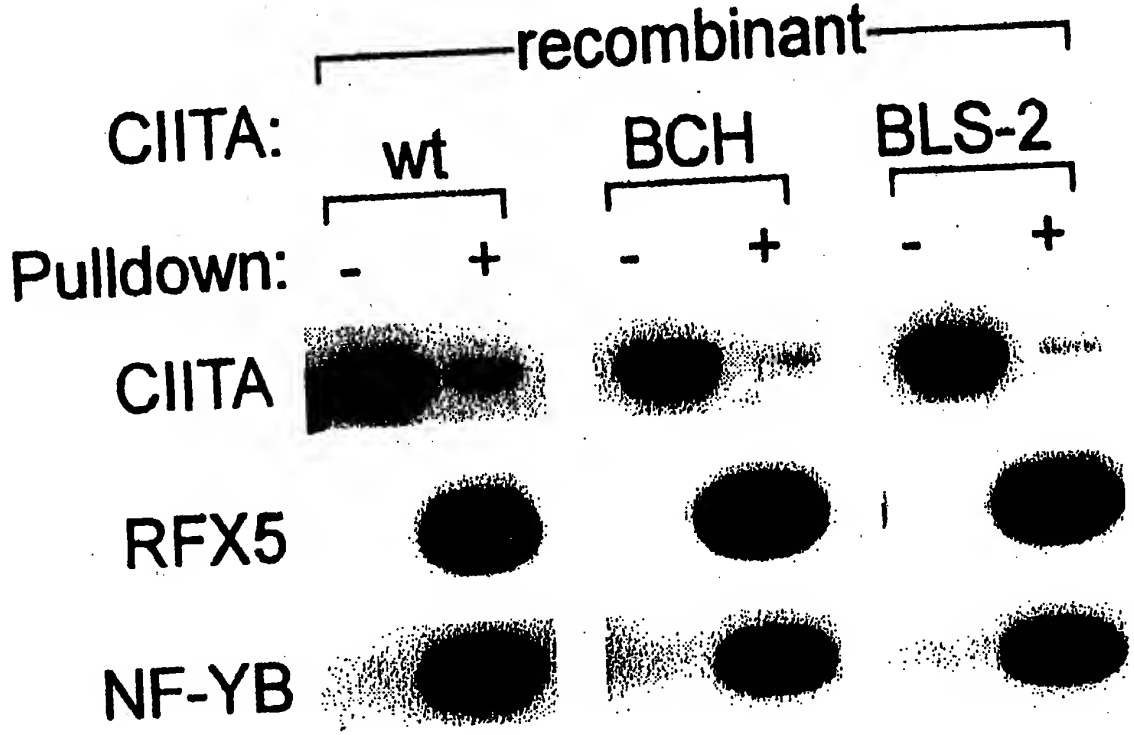


Figure 11d